

# Segmental spinal dysgenesis associated with occult dysraphism: Considerations on management strategies

## ABSTRACT

Segmental spinal dysgenesis is a rare and challenging entity especially when associated with occult dysraphism. Experience with a female patient followed during a period of 10 years spanning from 5 to 15 years of age is reported. During that period the girl underwent three spinal operations consisting in one decompression and spinal cord untethering, one posterior instrumented fusion and a spinal cord re-untethering. Clinical and radiological features are discussed and considerations on optimization of management strategies are made.

**Keywords:** Occult spinal dysraphism, segmental spinal dysgenesis, surgery, tethered cord

## INTRODUCTION

There is still a paucity of reports on segmental spinal dysgenesis (SSD), a rare congenital malformation resulting in aberrant segmentation and incomplete formation of the lumbar or thoracolumbar spine. Due to the ensuing structural deficit there is generally an association of kyphosis, stenosis and vertebral displacement of varying degree and malformation of the inferior limbs.

Because of this, before its recognition as a pathological entity in 1988,<sup>[1]</sup> various terms had been applied to this complex pathological picture, like “Congenital kyphosis and subluxation of the thoracolumbar spine due to vertebral aplasia”<sup>[2]</sup> or “Congenital spinal stenosis”<sup>[3]</sup> amongst others. Furthermore, in addition to the bony anomalies there is frequent association of intradural pathology like tethered cord and/or syringomyelia.

We describe our experience with a girl that has come to our observation at 8 years of age after having been operated on her spine already once elsewhere and who underwent two more operations at our department in the following 7 years.

## CASE REPORT

This girl was born in Guinea Bissau (Africa) and was brought to Italy at 5 years of age by a charity organization for medical treatment. At time of arrival orthostasis was not possible because of bilateral club foot and bilateral knee and hip dysplasia. A lumbar bony spur was palpable. She was treated for malaria, amebiasis, giardiasis and ancylostomatidae infestation at another hospital, where she also underwent urological investigation for incontinence and detrusorial hyperreflexia.

Imaging was performed at that same hospital and her computed tomography (CT) and magnetic resonance imaging (MRI) scans showed a complex lumbar malformation, with vestigial L3 and L4 vertebral bodies. These were

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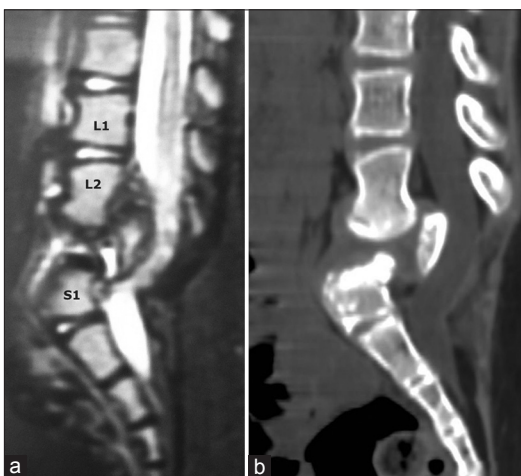
represented by hypoplastic posterior elements and an anterior bony remnant that retropulsed and compressed a low lying conus medullaris. Initial syringomyelia was also visible. At the inferior end a near to normal sacrum was present with what appeared to be an entirely sacralized L5 vertebra which was directly tagged as S1 [Figure 1]. The sacrum displayed a posterior schisis which together with the intradural finding completed the picture of an occult spinal dysraphism.

She therefore underwent a posterior decompression of the neural elements at the level of the segmental dysgenesis and a release of the tethered cord by intradural section of the filum terminalis.

She regained optimal bladder control after that surgery. In 2008 she underwent bilateral knee derotation with distal femoral osteotomy and fixation. Outcome was excellent and after intensive rehabilitation she started to walk with minimal assistance.

Two years later, in 2010 at 8 years of age, she once again lost bladder control, this time associated with excruciating pain irradiation in both lower limbs and progressive numbness in orthostasis. Standing and sitting became progressively more difficult and on sitting and any kind of forward flexion of the trunk a bony hump became visible in the lower lumbar region which disappeared upon extension. X-rays were thus made both in the sitting and recumbent position and a clear instability at the lumbosacral junction was visible with a retrolisthesis of the entire spine on the sacrum [Figure 2].

At this stage she was referred to our institution for further care.



**Figure 1:** (a) Sagittal T2 weighted magnetic resonance imaging showing the segmental dysgenesis. Note the vestigial remnant of the L3 and L4 vertebrae anteriorly compressing a low lying conus. Note also the incipient syringomyelia at D12-L1 level. (b) Sagittal computed tomography scan of the same section

When she came to our attention, still at 8 years of age, motor and sensory functions appeared intact, walking was not possible due to pain and there was no bladder control.

Due to the good reducibility of the spinal dislocation and an MRI that showed a good lumbar diameter in the reduced state in supine position we opted for an iliolumbar fixation with pedicle screws and rods in L1 and L2 and bilateral iliac screws. The patient underwent the operation without complications.

Postoperative X-rays showed correct positioning of the implants. The girl quickly regained full bladder control and motricity. She subsequently thrived for 6 years and was able to walk autonomously with the use of one or two crutches being able to cover short distances even without any aid. An X-ray done 2 years postoperative after a fall showed a broken left iliac screw with substantial stability of the system and the patient was asymptomatic [Figure 3].

At the age of 15, 6 years after the ileolumbar fixation and 4 years after the incidental finding of the broken iliac screw, the girl, who up to this moment was able to walk unassisted and had bladder control, experienced a sudden loss of urine, not accompanied by pain or any other complaint. In the following days to weeks these episodes increased in frequency and a progressive weakness in the lower limbs as well as increasing pain started to appear. This lead up to a condition were she was unable to sit or stand without excruciating pain in the lower limbs and the pain eased only in the supine position.

At this stage we were contacted and transferred the patient to our institution for assessment and treatment. Upon arrival motor examination showed a 2/5 motor deficit in both hip



**Figure 2:** (a) Lateral X-ray in flexion of the torso. Note the complete displacement of the vertebral column on the sacrum. (b) Lateral X-ray in extension showing realignment

flexors with an inability to lift the legs of the bed and a 3/5 deficit for the knee extensors. Sensation appeared to be intact also in the perineal area.

A CT scan showed maintained alignment of the lumbosacral junction with a solid fusion mass extending from L1 downwards but with an interruption at the S1 level. In addition to that the right iliac screw showed a windshield wiper loosening sign. An MRI scan revealed no new intradural findings with absence of compression of the neural elements and absence of syringomyelia [Figure 4].

Considering the “normal” intradural findings along with the interrupted fusion mass on CT scan, the left broken iliac screw and the right iliac screw loosening as well as the exacerbation of the symptoms upon flexion of the torso we were suspicious of pseudoarthrosis and motion at the lumbosacral junction and that this might have led to the clinical findings.

A passive flexion-extension lateral X-ray was thus performed which however failed to demonstrate any motion [Figure 5].

Considering all the information, in particular the insidious onset of symptoms with initial pain free loss of bladder control progressively leading to paraparesis, we opted for a diagnosis of tethered cord syndrome and decided to perform a surgery of intradural filum terminalis section.

Surgery was performed with removal of the residual L2 spinolaminar complex so as to obtain an untouched dural exposure. The dura was opened in a standard midline fashion and the filum terminalis was identified easily based on its distinct whitish and more structured aspect with respect to the rootlets of the cauda equina. No intraoperative monitoring was necessary. The filum, which did not appear to be under any particular tension, was thus coagulated and sectioned and the dura closed primarily in a watertight fashion.

Postoperative course was uneventful and after 2–3 days after the normal postoperative pain had settled a clear and progressive improvement became evident. Sitting position and flexion of the torso were possible again without pain and physiotherapy was started. The patient was discharged 8 days postoperative with hip flexors 3/5 and knee extensors 4/5. After intense physiotherapy in the following months she gradually regained the ability to walk. One year postoperative she is able to walk unassisted with the help of two crutches and needs to use diapers for persistence of reflex incontinence. Bowel function and sensation had returned to normal around 2 months postoperative.

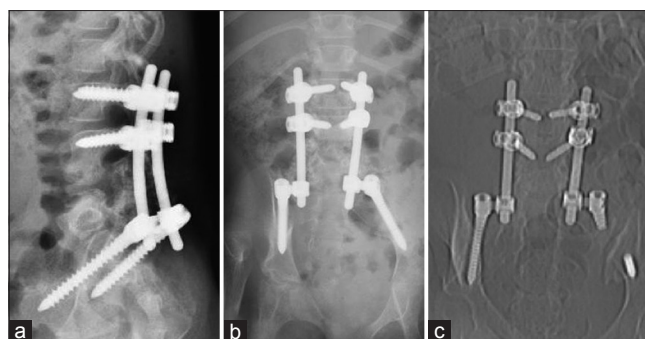


Figure 3: (a) Lateral X-ray showing instrumentation from L1 to iliac crests. (b) Anteroposterior X-ray of the same. (c) Anteroposterior computed tomography scout view showing breakage of the left iliac screw 6 years after implantation. Note the lateral displacement due to horizontal growth of the pelvis

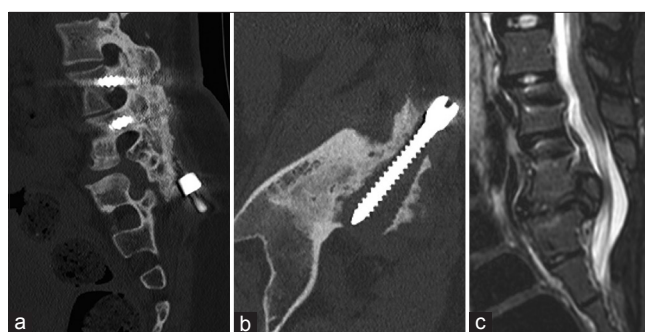


Figure 4: (a) Sagittal computed tomography scan showing fusion mass from L1 downwards. Note the interruption at the S1 articular facet. (b) Sagittal computed tomography scan showing windshield wiper loosening of the right iliac screw. (c) Sagittal T2 weighted magnetic resonance imaging showing maintenance of good decompression and absence of any relevant new intradural alterations

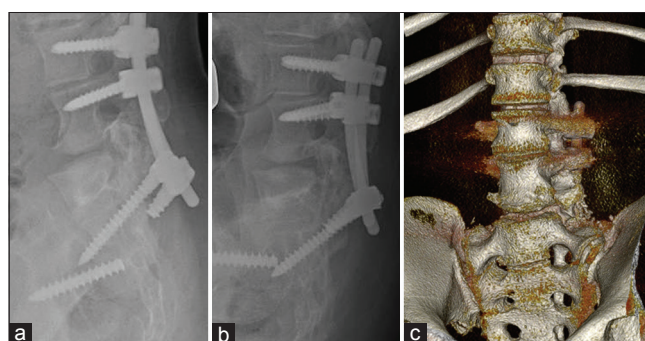


Figure 5: (a and b) Lateral X-ray in flexed and extended positions respectively. No apparent movement is detectable at the sacrolumbar junction. (c) Computed tomography three-dimensional reconstruction showing the anterior aspect of the thoracolumbosacral segment

## DISCUSSION

The complex and variable abnormalities in the SSD spectrum give rise to a difficulty and lack of formal classification. In our case we might be in front of an overlap between SSD and caudal regression syndrome, as the level of segmental

anomaly is too low for a further development of a spinal cord segment as beautifully explained by Tortori-Donati *et al.*<sup>[4]</sup>

Multistep surgery to correct associated malformations is a common feature, as these children have a wide spectrum of renal, bladder and articular abnormalities, and many have a severe motor impairment as early as newborns; however, even in those with some kind of motor sparing, spinal surgeons need to question themselves about a correct timing of surgery, which will be invariably needed at some point to correct the instable spinal situation, whether ultra-early with imaginable difficulties for bone to obtain fusion before 2–3 years of age, or later in time. Decompression is also discussed in children presenting with paraplegia, but assumed to be useful in those with motor sparing and evidence of superimposed damage due to curve progression in a stenotic canal. Dethetering may also improve progression of the curve.<sup>[5-7]</sup>

Our patient came to the first medical observation at 5 years of age after having avoided the risk to be literally “thrown into the sea” by the people of her village as this unfortunately still appears to be common practice with disomorphic and otherwise disabled children as it is interpreted as a divine curse. At the time of presentation motor function was still present and valid but bladder control was lost.

During that period she underwent her first spinal operation consisting in a simple posterior decompression and reported section of the filum terminalis. As a matter of fact she made a remarkable recovery after the surgery and regained both bladder control and the ability to walk after additional orthopedic interventions.

With hindsight it is surely appropriate to say that at that stage a fusion procedure should have been integrated in order to prevent the frank vertebral instability which 2 years down the line from that led to a new deterioration and loss of bladder control.

As to the most appropriate type of intervention for fusion this should have probably been an anterior procedure with bone on bone fusion after appropriate preparation between S1 and the lumbar spine with anterior plating and with or without an additional posterior procedure. We had decided to perform only a posterior instrumented fusion which, even though efficacious from a clinical point of view as well as from a radiological one (no instability was evident on dynamic X-ray), failed however to produce a complete fusion on CT scan. There surely is a solid fibrous union between the sacrum and the malformed lumbar end of the spine but we do not know how the lack of a continuous bony fusion mass will reflect itself on future development.

As to the intradural part of spinal cord untethering our choice to perform a second untethering procedure proved to be successful even though retethering in the absence of other associated malformations, as for instance lipomas, seems to be a rather rare event.<sup>[8]</sup>

This raises the question whether during the first operation of decompression an intradural section of the filum terminalis was actually really performed and if yes, whether that procedure was actually efficacious. Surely the second prepuberal growth spurt of the girl had led in the first instance to the asymptomatic breakage of the left iliac screw and later on to an increased tension on the spinal cord and thus manifestation of the tethered cord syndrome, but this was probably only possible because the cord had retethered or was probably never really untethered in the first place during the first surgery.

Finally, more frequent follow up assessments, in particular with regards to monitoring of the bladder function, might have helped to identify the ensuing last deterioration earlier with a more timely third operation and possible positive repercussions on bladder control.

## CONCLUSION

SSD spectrum remains a challenging decisional and surgical road. An early and multidisciplinary approach is mandatory for the best motor preservation and care of the patient. Fusion should be done early on and be circumferential and as short as possible in order to guarantee the best possible result at the first attempt and accommodate future growth of the spine without problems. Follow up should be particularly vigilant during growth spurt periods, especially in case of associated dysraphisms. Due to the rarity of this entity reporting and sharing knowledge of all cases is paramount to further optimize management strategies.

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## Conflicts of interest

There are no conflicts of interest.

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